

MHE Research Foundation

Multiple Hereditary Exostoses ("MHE") also often commonly referred to as Hereditary Multiple Exostoses ("HME")

Multiple Osteochondromas ("MO") is the preferred term used by the World Health Organization ("WHO")

Exostosis / Osteochondroma is a benign tumor that contains both bone and cartilage and usually occurs near the end of long bones. A Single Exostosis / Osteochondroma are one of the most common benign bone tumors found in children.

Multiple Exostoses / Osteochondromas is where people develop these tumors in multiple locations through out the body and has a prevalence of one in 50,000 people; however this is thought to be an underestimate as more mildly affected individuals may not come to the attention medical professionals in research studies.

Anatomical location	Percentage involvement
Distal femur	70 %
Proximal tibia	70 %
Proximal fibula	30 %
Proximal Humerus	50 %
Scapula	40 %
Ribs	40 %
Distal radius and ulna	30 %
Proximal femur	30 %
Phalanges	30 %
Distal fibula	25 %
Distal tibia	20 %
Bones of the foot	10-25 %



Regular Orthopaedic check ups are recommended to follow this condition. Check ups for children usually about every 6-9 months and adults every 24 months. Please remember when a physician tells you to make a follow up appointment, if the patient is having symptoms sooner they should contact there treating physician and be checked. Surgery is not required unless exostoses / osteochondromas become problematic.

The severity of MHE / MO/ HME varies from family to family and patient to patient. The number and size of exostoses / osteochondroma's vary widely. Exostoses / osteochondroma's can cause numerous problems, including: compression of peripheral nerves or blood vessels; irritation of tendons and muscles resulting in pain and loss of motion; skeletal deformity; short stature; limb length discrepancy; chronic pain and fatigue; early onset arthritis. General rule of thumb is that when an exostosis / osteochondroma is surgically removed that if it does not grow back within six months, then if sighted again this would be a new exostosis / osteochondroma.

MHE / MO / HME patients undergo surgical procedures to remove painful or deforming exostoses / osteochondroma's and or to correct limb length discrepancies and improve range of motion. Surgery, physical therapy and pain comprehensive management are currently the only options available to MHE / MO / HME patients.

What are the symptoms of problematic Exostoses / Osteochondroma?

The following are the most common symptoms of problematic exostoses / osteochondromas.

Keep in mind that each child, adolescent or adult can experience symptoms differently, depending on the size and location of the tumor or tumors:

- ◆ Exostoses / Osteochondromas are themselves a hard painless masses (becomes painful when they press or catch on tendons, muscles or nerves)
- ◆ Inflammation, Bursa formation and resulting bursitis may occur
- ◆ Irritation of a nearby nerve (weakness, numbness)
- ◆ Blood vessel aneurysm from pressure of the blood vessel by the Exostoses / Osteochondroma
- ◆ Angular deformity and Limb length discrepancy (Bowling of the limbs, Gait and Posture)
- ◆ Neurological problems (can also associated with spinal tumors)
- ◆ Loss in range of motion (ROM)
- ◆ Higher prevalence to scarring (surgery related)

How is an MHE / MO / HME (Exostoses / Osteochondroma) diagnosed and followed?

In addition to a taking a **complete medical history** and **performing a full physical examination**, your doctor may use one or more of the following tests to make the diagnoses or to follow the progression of this disorder.

X-rays - a diagnostic test that uses invisible electromagnetic energy beams to produce images of bones, onto film or digital image that can be viewed on a computer.

Magnetic resonance imaging (MRI) - a diagnostic procedure that uses a combination of large magnets, radiofrequencies, and a computer to produce detailed images of organs and structures within the body. This test is done to rule out any associated abnormalities of the spinal cord and nerves.

Computerized tomography scan (also called a CT or CAT scan) - a diagnostic imaging procedure that uses a combination of x-rays and computer technology to produce cross-sectional images (often called slices), both horizontally and vertically, of the body. A CT scan shows detailed images of any part of the body, including the bones, muscles, fat, and organs.

Bone scan - a nuclear imaging method to evaluate any degenerative and/or arthritic changes in the joints; to detect bone diseases and tumors; to determine the cause of bone pain or inflammation.

The malignant condition- associated with exostoses / osteochondromas is chondrosarcoma, a cancer of cartilage; there an increased risk of developing chondro- sarcoma reported risk of 2%-5% over ones life time.

Genetics of MHE / MO / HME- is an autosomal dominant genetic bone disorder. This means that a patient with MHE / MO / MHE has a 50% chance of transmitting the disorder to his / her children. This is equal for both male and female patients. There are two known genes that cause this disease EXT1 located on chromosome 8q23-q24 and EXT2 located on chromosome 11p11-p12. Approximately 60 to 70 % are located on the EXT1 gene and 20 to 30% are located on the EXT2 mutation. Most individuals with MHE / MO / HME have a parent who also has the condition, however, approximately 10% -20% of individuals with MHE / MO / HME have the condition as a result of a spontaneous mutation are thus the first person in their family to be affected. Clinical Genetic testing is available and can be obtained only by seeing a genetic counselor. Genetic testing results can be used for reproduction, **once the mutation is found** then **prenatal testing** and **Preimplantion Genetic Diagnosis can be offered by a genetic counselor.**

For more detailed information about MHE / MO / HME, guides on Orthopaedics, Chronic Pain, Genetics, Locating physicians, Research being conducted and other useful resources please visit our website www.mheresearchfoundation.org and or Please contact: Sarah Ziegler, National Director of Research and Coordinator of Clinical Information, Executive Director of the National MHE Research Registry.

All information located on the MHE Research Foundation website and printed materials has been written or reviewed by the Medical and Scientific Advisory Board.

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